Primary Carcinosarcoma of the Skin in an African Albino: Case Report and Review of Literature

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Abstract

Albinism results from deficiency in the protein responsible for melanin production in melanocytes. Albinos are at increased risk of dermatoses and malignancies. Primary carcinosarcoma of the skin is an exceeding rare biphasic tumour composed of malignant epithelial and mesenchymal elements with unclear histogenesis. To our knowledge, this has not been reported in an African albino. We report a case of this rare tumour in an African albino.

Keywords: Albinism, Carcinosarcoma, Immunohistochemistry, Skin cancer

Introduction

Albinism is a heterogeneous group of inheritable autosomal recessive disorders characterized by lack or defective tyrosinase enzyme which convert tyrosine to melanin precursor-dioxyphenylalanine, leading to the absence or reduction in melanin production in the melanocytes of the the skin, eyes, and hair.[1] It can occur in syndromic and non-syndromic

forms.[2] The global incidence of albinism is 1:20,000 individuals, with the prevalence in sub-Saharan Africa ranging from 1:15,000 to 1:1000.[2] According to the Albino foundation, there are about two million albinos in Nigeria.[3] The lack or dysfunctional state of melanin in the albinos exposes them to the deleterious effects of ultraviolet radiation (UVR), predisposing individuals to cutaneous and ocular pathologic conditions,[4] including skin cancers, commonly squamous cell carcinoma and basal cell carcinoma.[5]

Primary Carcinosarcoma of the skin (aka metaplastic carcinoma, biphasic sarcomatoid carcinoma, malignant mixed tumour) is an extremely rare biphasic tumour consist of an intimate admixture of epithelial and heterologous mesenchymal elements, both of which are malignant.[6,7] The histogenesis remains unclear. The most widely accepted theory is the monoclonal hypothesis, which postulates the divergence of a single totipotential stem cell into separate epithelial and mesenchymal directions.[8] Some however concluded that the sarcomatous component of the tumour is best regarded as a metaplastic transformation of the carcinomatous component.[6,9] Also, some authors have suggested the possibility of a collision of two different tumours or pseudo-sarcomatous reaction within the epithelial malignancy.[10] Only about 120 cases of cutaneous carcinosarcoma have been reported to date in the literature.[11] To our knowledge, it has not been reported in an African albino. Here we present a primary carcinosarcoma of the skin in a Nigerian albino.

Case report

A 57-year-old female albino patient of African ancestry presented with a year history of multiple skin lesions. She had an ulcerated tumour (5x5cm) on the nasal bridge extending to the medial canthal region (***Figure 1)***, a globular pedunculated growth (10x8x8cm) hanging from a stalk on the right side of the neck and ulcerated lesions on the right forearm and upper

back. There were no palpable lymph nodes. Review and examination of other systems were normal.

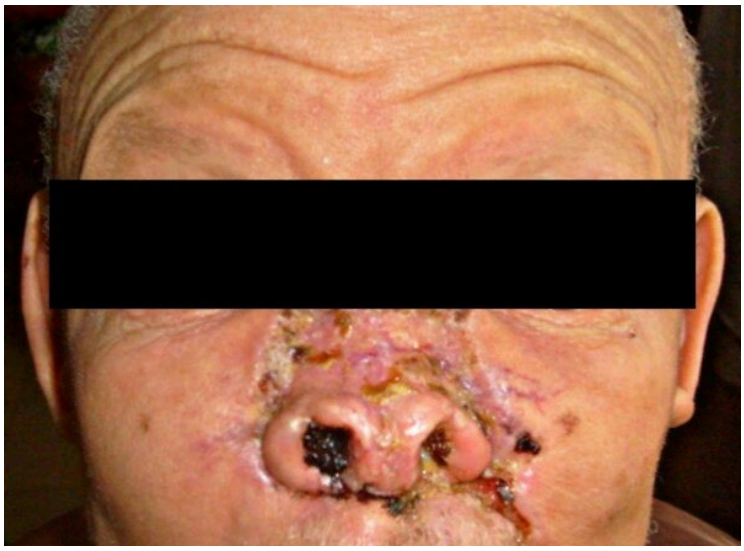

Figure 1: Shows the ulcerated tumour on the nasal bridge extending to the medial canthal region

Patient had a past history of wide local excision with tumour free margins and grafting for similar lesions diagnosed as squamous cell carcinoma about 3years prior to presentation. There is no past history of irradiation.

A chest x-ray and abdominopelvic ultrasound was done to rule out a visceral primary. Computed Tomography scan and Magnetic Resonance Imaging could not be done due to financial constraint. Ancillary laboratory investigations were essentially normal except for mild anaemia. Patient was admitted and had an excisional biopsy of the lesions. Wound care for the skin lesions following wide excision included alternate day wound dressing with sulfra-tulle gauze. No grafting done, and no recurrence noted one year post-excision.

Histology sections (***Figure 2***) showed a biphasic malignant tumour arising from the overlying epidermis and composed of atypical cells having hyperchromatic, vesicular nuclei and moderate cytoplasm. These cells are disposed in nests, trabaculae, cords and adenoid pattern and intimately admixed with malignant stroma having atypical pleomorphic spindle cells.

There are osteoid and cartilage formation within the malignant stroma. There is high mitotic activity with a count of 25/10hpf, including abnormal forms. There was no evidence of neural or vascular invasion. Immunohistochemistry results (***Figure 3***) show the epithelial component to be positive for pancytokeratin and p53, while the sarcomatous component is positive for Vimentin and p53. A diagnosis of Primary Carcinosarcoma of the Skin was made. Patient was followed up for six months post-surgery and had good outcome.

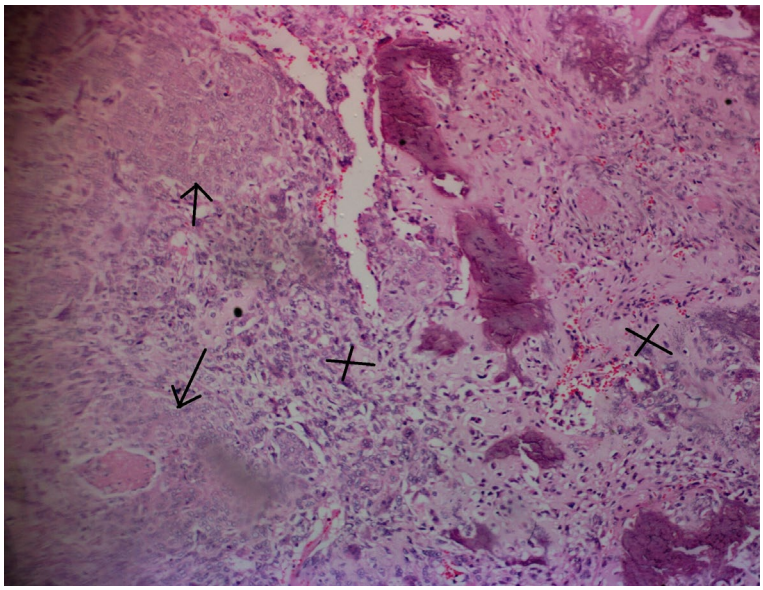

Figure 2: Histology sections showing a biphasic malignant tumour (H and E)

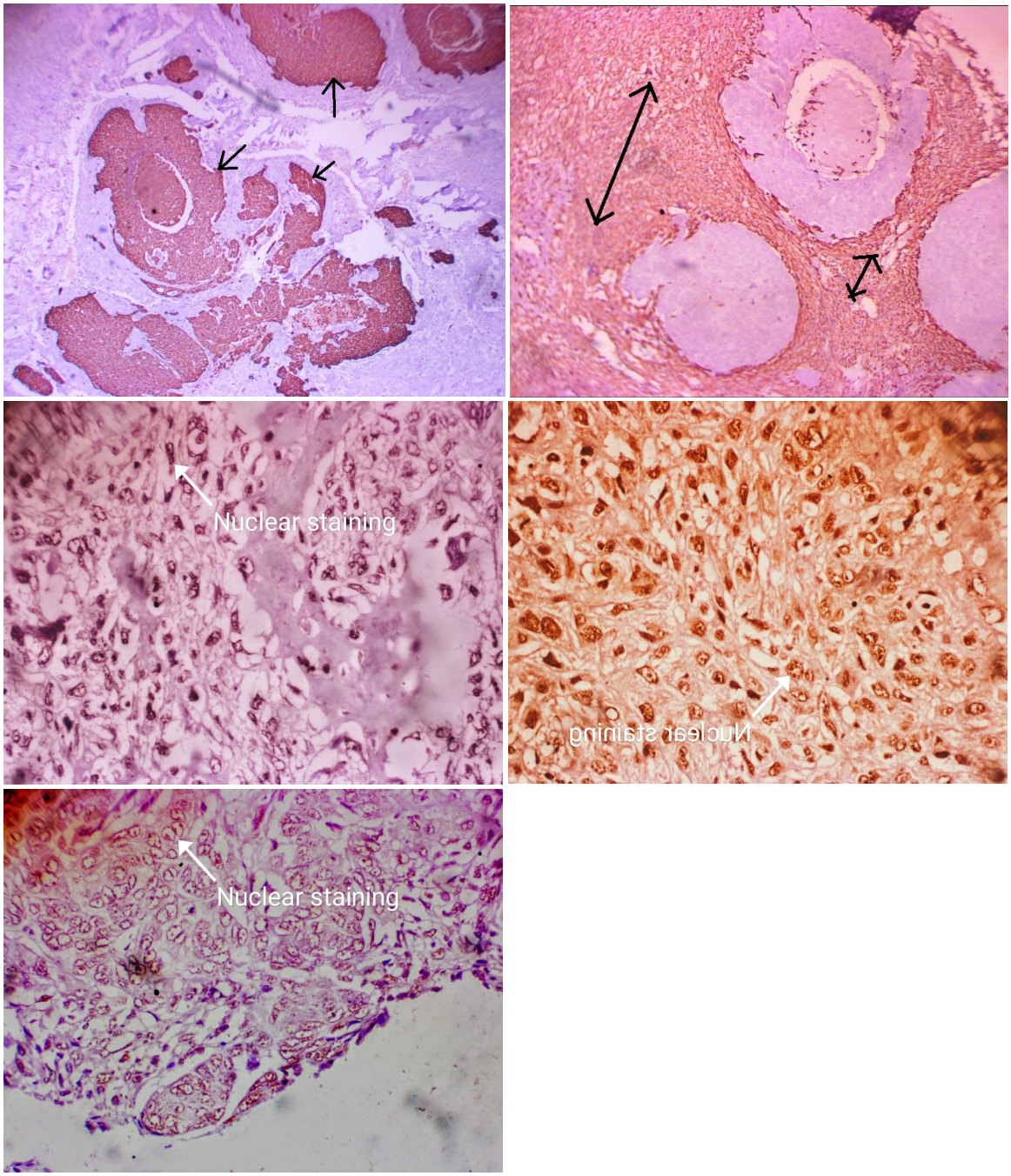

Figure 3: Immunohistochemistry show only the epithelial component to be positive for pancytokeratin (3a), only the sarcomatous component being positive for Vimentin (3b) and both components being positive for p53 (3c-3e)

Discussion

Skin cancer is rare among Africans and albinism is an established risk factor for skin cancer in this population.[12] Albinism is a genetically inherited disorder with a worldwide distribution. Phenotypically, it presents with reduced or no melanin in the hair, skin and eyes.

Ultraviolet radiation is highest at the equator and African albinos living close to the equator have the highest risk of developing skin cancers. Albinos develop malignant skin lesions at a younger age than the general population usually from the third decade of life.[12] The usual culprits are squamous cell carcinoma, basal cell carcinoma and malignant melanoma,[12] as well as basosquamous carcinoma and collision of squamous cell carcinoma and basal cell carcinoma.[13]

The index female patient was diagnosed of carcinosarcoma at age 57 years, earlier than the reported mean age of 71.5 years.[11] Case series by Kwak et al and Clark et al showed male preponderance.[11,14] Microscopically, the more common carcinoma component is a squamous cell carcinoma followed by basal cell carcinoma, whereas the most common sarcomatous component is osteosarcoma.[6] Other malignant heterologous mesenchymal elements include chondrosarcoma, leiomyosarcoma, rhabdomyosarcoma, fibrosarcoma, angiosarcoma, giant cell tumour of soft parts.[7] The index case is an admixture of squamous cell carcinoma and sarcoma with osteosarcomatous differentiation.

Cutaneous carcinosarcoma is broadly classified into two distinct groups.[6,15] Epidermal-derived (basal or squamous cell carcinoma epithelial component with a sarcomatous component) carcinosarcomas arise on the sun-damaged skin of the head and neck of elderly males (mean age 72 years) with a 70% 5-year disease-free survival.[6] In contrast, adnexal carcinosarcomas (spiradenocarcinoma, porocarcinoma, proliferating trichilemmal cystic carcinoma, or matrical carcinoma) present as recent growth in a long-standing nodule, are high risk as they occur in younger patients (mean age 58 years) and have a 25% 5-year disease-free survival.[6]

Immunoperoxidase stains are of great value in elucidating the components of this tumour. While the cytokeratin marker AE1/AE3 may be negative in the epithelial component, p63 and MNF116 are often expressed in poorly differentiated epithelial cells.[7] In cases with basaloid

epithelial component (basal cell carcinosarcomas), markers such as BerEP4 and Bcl-2 have also shown positivity.[16] In addition, p53 has been reported to be overexpressed by both epithelial and mesenchymal components.[16] However, the case series by Kwak *et al,* showed such co-expression in only two of the eleven cases, no expression in either component in one case while four cases each had p53 expression in either the epithelial or mesenchymal component.[11] The mesenchymal component may express vimentin, actin, CD10, CD34, and CD68, depending on its differentiation.[7] There was co-expression of p53 by the carcinomatous and sarcomatous elements in the index case.

Differential diagnoses include spindle cell squamous cell carcinoma and biphasic synovial sarcoma. Spindle cell squamous cell carcinoma consists of malignant spindle cells with variable components of invasive squamous cell carcinoma.[16] If spindle cells are not clearly segregated to epithelial or mesenchymal component with expression of cytokeratin with or without p63, they are confirmed as spindle cell squamous cell carcinoma rather than carcinosarcoma.[14,16] Up to 40% of spindle cell squamous cell carcinoma expresses vimentin. Therefore, it is difficult to use vimentin in carcinosarcoma for diagnostic purposes.[16,17] However, vimentin can provide additional information in differentiating the 2 neoplastic components of carcinosarcoma.[16,17] Biphasic synovial sarcoma consists of cord, nest, or gland- shaped epithelial cells and spindle cells with expression of TLE1 or EMA in both cell types.[14,16]

In contrast to metaplastic carcinomas arising in visceral sites, those primarily arising in the skin do not appear to behave in a very aggressive manner.[6,15] Complete excision of the lesion with 10mm-free circumferential margin is the treatment of choice; adjuvant chemotherapy or radiotherapy has not been shown to be beneficial.[8] The index case had wide local excision and has had no recurrence more than a year post excision.

In conclusion, we have presented a rare case of primary cutaneous carcinosarcoma in a Nigerian albino following recurrent cutaneous squamous cell carcinomas. Furthermore, we advocate increased enlightenment of albinos, especially those in Africa, on the use of sunscreens to prevent the incidence of skin malignancies among them.

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